Supplemental File S2: Contingency Tables

The following contingency tables were used to determine the significance of overlap for each Venn diagrams in Figures 3, 4, and 6.

For shared inferred GO-BP analyses, the background were the set of all GO BP terms that were inferred from any disease in CTD. For shared direct gene analyses, the background were all genes associated to any disease in CTD. The "phyper" function in R 3.2.1 was used to analyze the following 2x2 contingency tables using the hypergeometric distribution.

The "chi.test" function in R 3.2.1 was used to analyze the overlap among the inferred GO-BP terms for Bipolar Disorder, Chronic Kidney Failure and Diaphragmatic Hernia using Pearson's Chi-squared test as shown in the last table.

Diseases Analyzed: osteoporosis and breast cancer
Disease A: osteoporosis
Disease B: breast cancer

Shared Inferred GO-BP

	Disease A	Not Disease A	Total (Background)
Disease B	929	3,143	4,072
Not Disease B	287	6,251	6,538
Total (Background)	1,216	9,394	10,610

p-value < 0.001

Shared Direct Genes

	Disease A	Not Disease A	Total (Background)
Disease B	9	424	433
Not Disease B	57	6,252	6,309
Total (Background)	66	6,676	6,742

p-value = 0.009

Diseases Analyzed: leprosy and multiple myeloma
Disease A: leprosy
Disease B: multiple myeloma

Shared Inferred GO-BP

	Disease A	Not Disease A	Total (Background)
Disease B	88	697	785
Not Disease B	180	9,645	9,825
Total (Background)	268	10,342	10,610

p-value < 0.001

	Disease A	Not Disease A	Total (Background)
Disease B	0	31	31
Not Disease B	12	6,699	6,711
Total (Background)	12	6,730	6,742

p-value = 0.054

Diseases Analyzed: angina pectoris and erectile dysfunction
Disease A: angina pectoris
Disease B: erectile dysfunction

Shared Inferred GO-BP

	Disease A	Not Disease A	Total (Background)
Disease B	110	430	540
Not Disease B	268	9,802	10,070
Total (Background)	378	10,232	10,610

p-value < 0.001

Shared Direct Genes

	Disease A	Not Disease A	Total (Background)
Disease B	0	10	10
Not Disease B	34	6,698	6,732
Total (Background)	34	6,708	6,742

p-value = 0.049

Diseases Analyzed: type 2 diabetes and Alzheimer disease
Disease A: type 2 diabetes
Disease B: Alzheimer disease

Shared Inferred GO-BP

	Disease A	Not Disease A	Total (Background)
Disease B	1,108	703	1,811
Not Disease B	1,189	7,610	8,799
Total (Background)	2,297	8,313	10,610

p-value < 0.001

	Disease A	Not Disease A	Total (Background)
Disease B	11	84	95
Not Disease B	122	6,525	6,647
Total (Background)	133	6,609	6,742

p-value < 0.001

Diseases Analyzed: ulcerative colitis and coronary artery disease
Disease A: ulcerative colitis
Disease B: coronary artery disease

Shared Inferred GO-BP

	Disease A	Not Disease A	Total (Background)
Disease B	398	644	1,042
Not Disease B	676	8,892	9,568
Total (Background)	1,074	9,536	10,610

p-value < 0.001

Shared Direct Genes

	Disease A	Not Disease A	Total (Background)
Disease B	0	60	60
Not Disease B	60	6,622	6,682
Total (Background)	60	6,682	6,742

p-value = 0.416

Diseases Analyzed: Bipolar Disorder, Chronic Kidney Failure and Diaphragmatic Hernia
Disease A: Bipolar Disorder
Disease B: Chronic Kidney Failure
Disease C: Diaphragmatic Hernia

Shared Inferred GO-BP

	Disease A&B	Disease A & Not Disease B	Disease B & Not Disease A	Not Disease A & Not Disease B	Total (Background)
Disease C	231	86	452	627	1,396
No Disease C	71	403	512	8,228	9,214
Total (Background)	302	489	964	8,855	10,610

Chi-Square = 2314.6, p < 0.001